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Brief report

Isolated intraspinal juvenile xanthogranuloma in an infant presenting as acute paraplegia

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Abstract

Juvenile xanthogranuloma is a histiocytic proliferative disease which predominantly affects the skin. Extracutaneous involvement is rare. We present a 6 month old infant with an acute paraplegia. MR-imaging showed an intraspinal extradural mass at mid-thoracic level with marked compression of the myelon. Complete tumor removal was achieved by emergency surgery, followed by complete neurological recovery. Histological examination led to the diagnosis of a juvenile xanthogranuloma. To our best knowledge, an isolated intraspinal juvenile xanthogranuloma in the first 12 months of life has not been described before.

INTRODUCTION

Juvenile xanthogranuloma was first described in 1905 by Adamson as a “congenital xanthoma multiplex” [1]. It is a benign, non-Langerhans histiocytic proliferative disorder of the skin which mainly occurs in childhood. In 80-90 percent it presents as a solitary cutaneous lesion [2,3,4,5,6]. Dermal juvenile xanthogranuloma is histopathologically characterized by mononuclear cells, multinucleated cells with or without Touton features and spindle cells [2]. In a study of Dehner, Touton giant cells were less common in extracutaneous lesions [4]. The immunhistochemical findings are important to distinguish from other Langerhans-cell and non-Langerhans-cell histiocytosis. Typically juvenile xanthogranuloma shows a CD1-negative and C68-positive histiocytic proliferation [2,5,7]. Etiology is unknown, it is believed that a granulomatous reaction results from a disordered macrophage response to a non-specific tissue injury [7]. In a study from Janssen and Harms with 129 patients male/ female ratio was 1.4:1, mean age at diagnosis was 22.4 months [5]. Spontaneous regression over years is common [4,7,9]. Some authors describe an association of juvenile xanthogranuloma and neurofibromatosis type I and juvenile chronic myelomonocytic leukemia [4,10].

In this brief report we present a 6 month old infant with an acute paraplegia caused by an isolated intraspinal juvenile xanthogranuloma which is exceptional.

CASE REPORT

The mother of this 6 month old infant initially noticed reduced activity of the lower limbs which worsened over 3 days, lifting of the legs was painful for the child. Bladder and bowel function was always inconspicuous.

On clinical examination we found a paraplegic baby, with no spontaneous movements of the legs, exaggerated deep tendon reflexes and positive Babinski sign. No pathological cutaneous findings were revealed. Immediately performed MR-imaging of the spine showed an intraspinal, extradural, extramedullar and spotted

contrast medium enhancing mass at level thoracic vertebra 3 to 7, with marked compression of the myelon [Figure 1, 2].

In emergency surgery (5 hours after first patient contact) laminoplasty T 3-T 7 and complete resection of the tumor was achieved as confirmed by postoperative MRI. Histological and immunohistochemical examination showed typically CD1-negative, C68-positive histiocytic proliferation which led to the diagnosis of a juvenile xanthogranuloma. Touton giant cells could not be detected. The infant showed full clinical recovery. In the subsequent 2 years follow up the child hat no neurological deficits and MR-neuroimaging (in 1y and 2y follow up) remained normal.

DISCUSSION

Intraspinal tumors are a histologically heterogenous group of rare tumors. In the pediatric age group extradural intraspinal tumors account for about 50% of cases and are likely to be neuroblastomas and Ewing's sarcomas, and more rarely, leukemia, lymphoma, rhabdomyosarcoma or – as in our case – juvenile xanthogranuloma.

Extracutaneous involvement of juvenile xanthogranuloma is with 5-10% rare, mostly affected are central nervous system, eyes, liver, spleen, lungs and kidneys [2,3,4,6] either as a systemic appearance where skin and several organs are involved or, as in our case, as a single parenchymal manifestation [4,5,9]. An isolated intraspinal juvenile xanthogranuloma in infancy and adulthood is exceptional, over all age groups, 8 cases of solitary juvenile xanthogranulomas involving the spine are reported until now [2,3,7,8,11,12,13,14], 5 patients presented with incomplete or complete paraplegia/ tetraplegia [3,11,12,13,14]. 1993 first intraspinal xanthogranuloma in an 13 month old infant was reported by Shimosawa et al. [14], in the pediatric population up to now 6 cases are described [3,7,8,12,13,14]. In 2 cases intraspinal xanthogranuloma was located in the cervical spine [7,13], in 2 cases thoracal spine was involved [12,14]. 2/3 of the reported children presented, as our case, with incomplete or complete paraplegia/ tetraplegia.
In spinal MR scan juvenile xanthogranuloma appears in a homogenous signal pattern, in T1-weighted imaging mostly as a iso- or hypointense lesion. Differential diagnosis include various neoplasms like fibroma, fibrous meningeoma and solitary

fibrous tumors [2]. To approve diagnosis, histopathological and immunhistochemical methods are gold standard [2].

Cutaneous lesions have the possibility for spontaneous regression, in symptomatic lesions surgical excision is recommended [6]. Total removal seems to be curative and is recommended because of the high potential risk of recurrence [2,5,7,13,14]. If resection is not possible or systemic juvenile xanthogranuloma is present, radiotherapy and/ or multiagent chemotherapy may be instituted [4,6,9,14].

An isolated intraspinal juvenile xanthogranuloma in the first 12 months of life has not been described yet. No long term follow-up of intraspinal juvenile xanthogranuloma has been reported in the literature so far. Fulkerson et al. described a child with a solitary intracerebral juvenile xanthogranuloma with a 3-year follow-up period [15], like our patient the child remained asymptomatic since surgery. Immediate surgical decompression in our infant was essential for the complete neurological recovery.

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Figure legends

Figure 1: Sagittal T2-weighted (A) and T1-weighted MRI (B) show an intraspinal, extradural and extramedullar tumor at level thoracic vertebra 3 to 7.

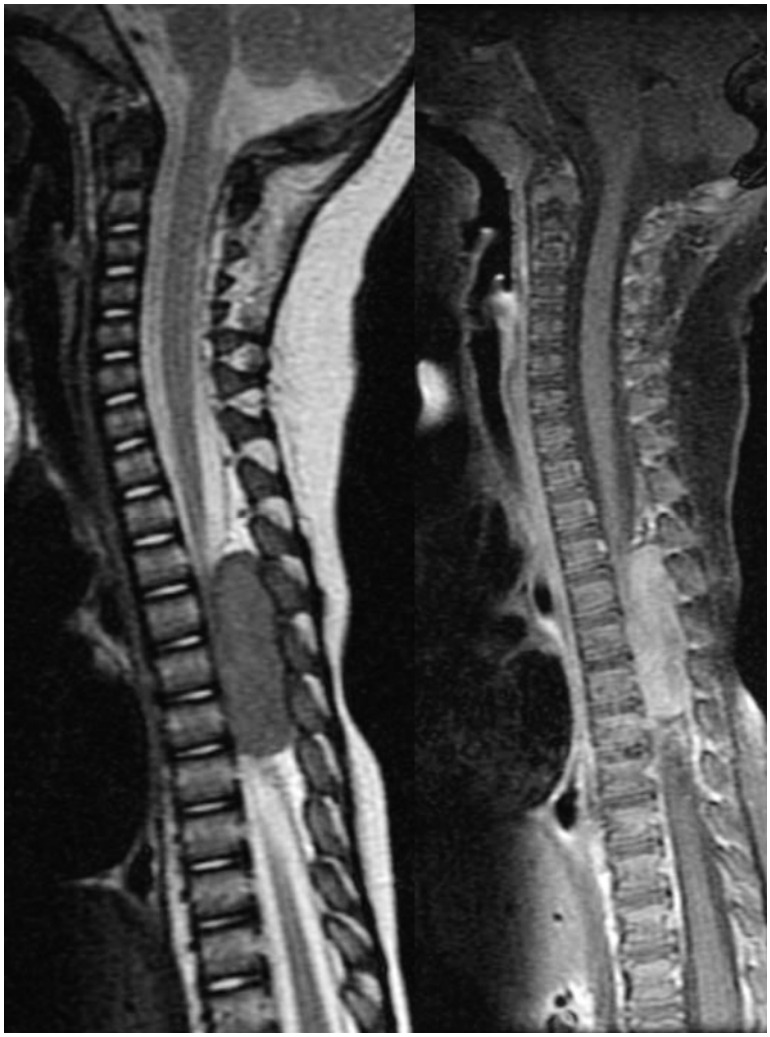

Figure 2: Axial T1-weighted MRI at level T3/ T4 shows a marked compression of the myelon (asterisk).